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Medical management of sensorineural hearing loss

PART II: Musculo-skeletal system

By J. B. Booth (London)

It is obvious that, by definition, the temporal bone must form part of the skeletal system but by contrast, whilst it may be affected by many specific conditions, it is relatively infrequently involved in many of the generalized systemic diseases of bone. No single classification is universally acceptable and the titles of many conditions are subject to change and reclassification.

Here, it is proposed to deal with only the following

Osteoporoses
- Primary
- Secondary
  - Acromegaly

Osteomalacias
- Hypophosphataemic (Vit. D)
  - Rickets and Osteomalacia

Hypercalcaemia
- Primary Hyperparathyroidism
- Other Hypercalcaemias
- Vit. D poisoning

Systemic Bone Disease
- (see Table I)

Conditions 1, 3, 6, 8 and 9 have intentionally been omitted from the discussion—1 and 3 because they are already well-known to otologists. 6, 8 and 9 because of their great rarity and because they are usually seen only in infancy.

The biochemical, radiological and other characteristics of those discussed are summarized in Table II.

TABLE I
SYSTEMIC BONE DISEASE

1. Otosclerosis
2. *Osteitis Deformans (Paget's disease)*
3. Osteogenesis Imperfecta (Van der Hoeve Syndrome)
4. Fibrous Dysplasia
5. Osteopetrosis
6. Genetic Craniotabular Dysplasias
   (a) Craniometaphyseal dysplasia
   (b) Frontometaphyseal dysplasia
7. Genetic Craniotabular Hyperostoses
   (a) Hyperostosis corticalis generalisata
   (b) Sclerosteosis
   (c) Congenital hyperphosphatasia
   (d) Progressive diaphyseal dysplasia
8. Craniofacial Dystosis
9. Osteopathia Striata (with Sclerotic Skull Base)
10. Neurofibromatosis

Osteoporosis

Henkin *et al.* (1972) described the otological findings in seven patients with idiopathic osteoporosis, five of whom had sensorineural deafness greater than would be expected for their age. The patients presented because of the severity of their bone pain, and were admitted for evaluation and treatment. All showed clinical features of the condition; fractures of thoracic and/or lumbar vertebrae; normal serum calcium, phosphorus, and alkaline phosphatase. Five of the seven patients were subjectively aware of their hearing loss which occurred concomitantly with or after the onset of symptoms of the bone disease. Radiology of the temporal bones showed increased sclerosis of cochlear and vestibular labyrinths in four of the seven patients. The hearing loss was gradual, with progressive impairment, and bilateral in all but one. Acoustic impedance measurements showed a low, normal compliance and the acoustic reflexes were present in all but one.

Acromegaly

Acromegaly is a chronic disease of middle life resulting from excessive secretion of growth hormone by the acidophil cells of the anterior pituitary.

Recently Graham and Brackmann (1978) reported three cases whose acromegaly had involved the temporal bone. Radiology demonstrated massive thickening of the mastoid cortex and posterior bony canal wall with secondary lengthening of the bony external meatus; some overgrowth diminishing the lumen may also occur. However the internal meatus, cochlea and vestibule appeared normal and the structures of the otic capsule, including the facial nerve, remained in normal relationship.

Earlier Richards (1968) investigated 15 patients (30 ears), including four who had developed diabetes (2 mild, 2 severe); five ears showed a

marked conductive deafness, but otherwise the remaining ears developed a sensorineural loss which was substantially lower than in the normal population, with a general tendency for the hearing loss to deteriorate with age, but there was no relationship between the duration of the disease and the deafness. He was unable to demonstrate any direct relationship between the plasma growth hormone levels and the hearing loss, and there was the suggestion that there might be an inverse relationship, i.e. the patients with the better hearing levels were those with higher hormone levels. (The caloric reaction was normal in all patients.)

Doig and Gatehouse (1981) assessed the hearing in 48 patients with acromegaly (including 8 with diabetes) and compared them with matched controls, but were unable to find any significant difference between the two groups nor any correlation with diabetes, growth hormone levels, blood pressure or other factors, and no change in the hearing occurred after surgery to remove the tumour.

Williams (1981—personal communication) noted that, in 150 acromegalic patients, no specific relationship could be found with the level of growth hormone and that the instance of otosclerosis remained the same as in the normal population.

Osteomalacia

(Hypophosphataemic Rickets—Vitamin D Resistant)

Hitherto nutrition rickets or osteomalacia have not been reported as a cause of deafness. However, there is evidence of continuing admissions of patients with these two conditions in the Asian community, particularly in such cities as Glasgow and Bradford. During the period 1968 to 1978 no fewer than 138 Asians were discharged from Glasgow Hospitals for these conditions and during the same period the Asian population of the city is estimated to have increased from about 8,000 to 14,000. During the same period the probability of an Asian schoolchild being admitted to a Glasgow Hospital with rickets was 1 in 29 and 1 in 7 (14 per cent) showed radiological evidence of active rickets; 2 out of 5 (40 per cent) had biochemical evidence, while 44 per cent had reduced serum 25-OHD concentrations). Mothers of three families had to be treated in hospital for severe osteomalacia (Dunnigan *et al.*, 1981).

A survey in April 1973 of schoolchildren in Bradford, the city with the largest Asian community in the United Kingdom, showed biochemical evidence of rickets to be present in 45 per cent of the Asian children. There it was estimated that 1 Asian child in 40 would require admission before adolescence for vitamin D deficiency. In the 1971 census there were some 652,000 Indian and Pakistani immigrant residents, of whom 16 per cent were under the age of 5, 35 per cent under the age of 15, and it would seem therefore that nutritionally induced osteomalacia in the adult community might also be expected to occur.

For the majority of deeply pigmented people in Britain, any effect of

TABLE II

Paget's Disease (Osteitis Deformans)	Conductive and/or Sensorineural	Ca usually normal PO_4 Hypercalcaemia—immobilization Alk -ase *elevated in active* disease Acid -ase may be raised Urinary HDP elevated in active disease	Varied—lytic, sclerotic and mixed phases Skull—great increase in thickness of both tables, particularly outer ? patchy sclerosis—wooly appearance Platybasia; Basilar impression	X-ray pelvis inc. Femoral heads Osteoporosis Circumscripta - patch of reduced density resembling bony defect Pathological Fracture
Fibrous Dysplasia	Conductive Rarely Sensori-neural	Ca usually normal PO_4 always normal Alk -ase may be raised in active disease, especially *poly*-ostotic form Acid ase normal	Monostotic/Polyostotic appearances—same Multiloculated cystic lesion (bone frequently expanded) *Occ. lesion more diffuse* ground-glass appearance due to multiple fine trabeculae Occ. diffuse sclerotic appearance	Skeletal Survey to exclude polyostotic Pathological Fracture Cafe au-lait pigmentation *may be present (either type)*
Osteopetrosis (a) Albers Schönberg	Conductive Occ. Mixed	Ca, PO_4 } normal Alk, Acid } -ase may be markedly elevated Urinary HDP usually normal	Symmetrical increase in bone density; bones appear structureless Sclerotic foci—'bones within bones' Thickening of Vertebral end-plates ('Rugger Jersey')	Thick dense brittle bones Pathological Fracture Facial Palsy Occ. osteomyelitis of mandible after dental extraction Mild anaemia
(b) Malignant Recessive	Sensorineural	As above	Transverse bands in metaphyseal regions of *long bones and longitudinal* striations Prox. humerus and distal femur—flask-shaped Vertebrae—rugger jersey	Facial Palsy Blindness Pathological Fracture Mental Retardation Liver and Spleen enlargement Haemolytic anaemia and Thrombocytopaenia

Genetic Craniotabular Hyperostoses				
(a) van Buchem's (Autosomal Recessive)	Conductive and/or Sensorineural	Ca, PO_4 } normal Alk -ase frequently raised (50–250 per cent)	Diffuse, symmetrical increase in bone density Cortical bone—abnormally thick but bones not increased in size Hyperplasia diaphysis long and short bones Endosteal thickening diaphysis —tubular bones	Normal stature Facial Palsy Clavicles—thickened and palpable Overgrowth of brow and mandible
(b) Sclerosteosis (Autosomal Recessive)	Conductive and/or Sensorineural	Ca, PO_4 } normal Alk -ase markedly elevated in nearly all patients Acid -ase normal	Bones show increased density but only minor degree of bony modelling, if present Progressive bony thickening Tubular bones markedly undermodelled with lack of usual diaphyseal constriction	Syndactyly and Digital Malformation Facial paralysis Tall stature Distortion of Face and Jaw Chronic headache Raised ICP Anosmia Majority Afrikaners
(c) Congenital Hyperphostasia (Autosomal Recessive)	Conductive with Decreased Bone Conduction	Ca, PO_4 } normal Alk, Acid } -ase both consistently elevated Urine-HDP high	Similar to Paget's Marked irregular thickening of skull ? Narrowing of EAM Tubular bones—width greatly increased, bowing and lack of modelling	Multiple Fractures Dwarfing Blue Sclerae (? increased serum uric acid and leucine aminopeptidase)
(d) Progressive Diaphyseal Dysplasia (Autosomal Dominant)	Combined with big Air-Bone gap	Ca, PO_4, Alk -ase, Acid -ase } usually normal	Generalized sclerosis of skull base; vault less commonly/severely affected	Marked thickening of cortices of leg bones medullary canals narrowed; external bony contours irregular

skin colour on their health is insignificant, but when there is a higher risk of rickets or osteomalacia for other reasons, a dark skin may add to the total risk and predispose to the development of overt disease. It is also known that phytate, or inositol hexaphosphate, which occurs in whole cereal grains, interferes with the absorption of calcium by forming insoluble calcium phytate in the intestine. A high fibre intake might also, under certain conditions, reduce the availability of calcium in the diet. If yeast is not added, then much less phytate is destroyed compared with normal breadmaking, when the dough is set to rise (*Rickets and Osteomalacia*, HMSO 1980).

Brooks and Morrison (1981) have described two cases in which vitamin D deficiency resulted in biochemical osteomalacia, and was associated with bilateral cochlear deafness. Both patients showed trough shaped fully recruited cochlear deafness, and tomography in one demonstrated bilateral cochlear demineralization (Fig. 1). After commencing calciferol replacement therapy (3,000 units daily), this patient showed a 12 db. mean unilateral hearing improvement two months later. (A follow up on the second patient was not possible.)

FIG. 1
Osteomalacia. Bilateral cochlear demineralization; arrow indicates obliteration of lumen of basal turn.

They noted that demineralization of the otic capsule may lead to secondary degenerative changes in the spiral ligament, stria vascularis and cochlear hair cells. They suggested a correlation between the vitamin D deficiency and cochlear deafness which has previously been unrecognized.

Heritable primary hypophosphataemia due to an isolated renal tubular leak of phosphate is the most common type of metabolic (vitamin D-resistant) rickets and is nearly always transmitted by an X-linked dominant gene (Fig. 2). Sporadic cases due to new mutation are not uncommon and such patients may be expected to transmit the disease to their offspring in the X-linked manner.

Stamp and Baker (1976) described two children out of a family of three by a first cousin marriage, who presented with severe rickets in infancy. The disease showed continued activity, marked resistance to treatment with vitamin D, early fusion of cranial sutures, greatly increased bone density,

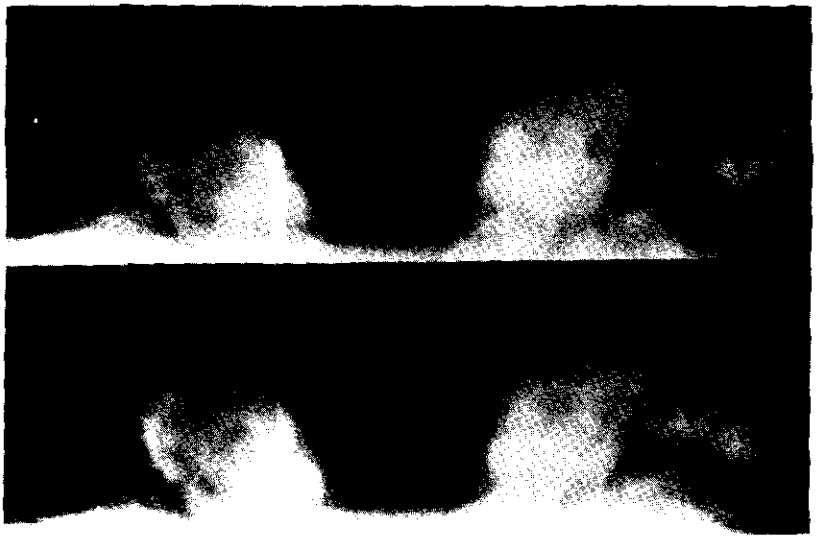

FIG. 2
Hypophosphataemic rickets. Sclerotic bone involving the whole of the petrous pyramid with overall expansion.

nerve deafness, and lifelong hypophosphataemia, unaffected by treatment. Tomography of the internal auditory canal showed marked narrowing at several points throughout its length.

In these patients there was an unexplained tubular phosphate leak similar to that occurring in X-linked rickets, and persisting despite adequate control of rickets with high doses of vitamin D. They considered the gross bony growth in their patients might have been the cause of the perceptive deafness. There is strong evidence of mild secondary hyperparathyroidism in patients with primary hypophosphataemia.

Weir (1977) described two pairs of siblings who were known to suffer from a recessive hypophosphataemic rickets, three out of four of whom developed some degree of sensorineural deafness, and all demonstrated positive radiological findings of marked narrowing of the internal auditory canals. In the X-linked hypophosphataemic variety, alkaline phosphatase levels returned to normal on cessation of growth, but in the recessive form continued biochemical activity persists on attaining adult stature, and maintenance therapy with vitamin D is necessary. The increase bone density cannot be attributed to the long-term high doses of vitamin D, but may be due to a mild secondary hyperparathyroidism.

Osteosclerosis in primary hyperparathyroidism

It should be added that osteosclerosis is common in renal osteodystrophy in which secondary hyperparathyroidism is almost universal. Osteosclerosis in primary hyperparathyroidism is extremely rare.

Hypercalcaemia

Primary hyperparathyroidism

This is rarely seen affecting the ear. Ruedi (1968) describes the temporal bone changes in two patients, and Lindsay and Suga (1976) in another. Involvement of the otic capsule would only seem to occur when the condition has reached the stage of 'osteitis fibrosa cystica' (von Recklinghausen's disease of bone). Morrison (1979) gives the details of one case—a 46-year-old man who presented with a one-month history of a rapidly progressive bilateral hearing loss.

Although many patients give evidence of excessive effects of parathyroid hormone on bone, only a few develop clinically evident osteitis fibrosa cystica. For some reason, which is not understood, the incidence of osteitis fibrosa cystica is on the decline.

Vitamin D poisoning

Cohen *et al.* (1979) reported a patient with pseudo-hyperparathyroidism, who continued to take calciferol 2.5 mgm. daily, and returned four years later with a three-month history of deafness. Examination showed an extensive calcification of the tympanic membranes and cornea, with a severe conductive deafness. Extensive calcification of the kidneys and blood vessels (renal function tests normal) was seen; X-rays of the mastoids showed these to be cellular. Despite treatment the deafness remained.

Paget's disease

There can be few conditions bearing an eponym which are so well known and which have provided such persistent interest. Sir James Paget described the condition which bears his name in 1877. It has an unusual racial and geographic distribution, being very common in the United Kingdom, Australia and New Zealand and in other populations of British origin, such as North America and South Africa. It has, therefore, been thought that it was a disease of Anglo-Saxon origin but there are now well-documented instances occurring in African and Asian subjects. There is no doubt the disease is common and it has been estimated that some 3 per cent of the population of England and Wales over the age of 40 may suffer from it; of these only a small proportion have symptoms or severe deformity, but it is thought that there may be almost 50,000 in Britain who have symptomatic Paget's disease.

Surveys have revealed a marked geographical variation in the disease and within the United Kingdom the prevalence has been shown to be considerably higher in Lancashire than elsewhere, but decreasing from high to lower levels over very short distances (Barker *et al.*, 1980).

Paget himself described the onset of the disease in middle age with slow progression which produces effect by changing the shape, size and

direction of the diseased bones. He noted the common enlargement of the skull, which nevertheless did not seem to cause brain compression, and drew attention to the very frequent involvement of the femora and tibiae (Davies, 1968).

Of those with widespread active disease, bone pain is a troublesome symptom, and probably occurs in as many as 20 per cent, sufficient to warrant treatment. Expansion of bones around foramina at the base of the skull and in the orbit can lead to neurological defects and optic atrophy.

The aetiology of the disease is unknown but pathologically it arises from an increase in the number and activity of osteoclasts. The stimulus for this increase is unknown, but as a result of the bone resorption, there is a compensatory increase in bone formation, leading to a very great increase in bone turnover, and the normal lamellar structure of the collagen of the ground substance becomes grossly disordered. The most interesting recent hypothesis is that the disease results from a slow virus infection of the osteoclasts. The pelvis is the area most commonly affected by the disease (76 per cent) and the skull is involved in 28 per cent.

More recently, Harner *et al.* (1978) reviewed the records of 463 patients with Paget's disease and noted that, while sensorineural loss was the most frequent type, it was usually not part of the disease process, and most patients had no evidence of temporal bone involvement. Their patients were seen from a total of 1,066 patients who had been seen with objective evidence of Paget's disease over a 5-year period. In their series, 17 per cent of patients had tinnitus and 21 per cent had dizziness; the most common type of the latter was postural positional unsteadiness. In those patients whose initial audiogram showed the mixed hearing loss the mean age was 56 years, and 49 per cent showed evidence of the disease before the age of 60. In those with a sensorineural loss the mean age was 61 years, with 37 per cent below the age of 60. In those in whom there was radiological evidence of skull involvement, the incidence of mixed hearing loss was statistically much greater than expected, and the incidence of tinnitus and dizziness was also higher than in the overall group.

Whilst an increased tortuosity and hypertrophy of the anterior terminal branch of the superficial temporal artery may be seen in many patients with skull involvement in Paget's disease, it is by no means characteristic of the condition.

X-ray. In the early stages small areas of lucency and dense patches are seen which fade into one another. There is often a typical mixture of lytic and sclerotic areas and the skull is thick where it is affected, predominantly over the vertex. Some coarse trabeculae are nearly always visible, except in the most advanced cases. Osteoporosis circumscripta is a different manifestation of the disease causing a total radiographic disappearance of bone, but always stopping short of involving the whole (Fig. 3) (Kasabach and Gutman, 1937; Du Boulay, 1980).

The tomographic changes in the temporal bone from Paget's disease

FIG. 3
Osteoporosis circumscripta (Paget's disease).

have been described by Petasnick (1969); these varied from minimal demineralization of the petrous apex to demineralization of the entire petrous pyramid, including the otic capsule, and they appeared to correlate grossly with the degree of skull involvement (Fig. 4). The involvement of the internal auditory canals consisted of demineralization of the walls without evidence of narrowing. In patients with extensive involvement, the internal auditory canal was no longer identifiable as a distinct structure; the margins of the external auditory canal and middle ear showed demineralization in those with extensive involvement. 28 of the 31 ears with sensorineural deafness had cochlear radiographic changes.

The medial aspect of the petrous pyramid is the initial site of involvement, followed by progressive involvement of the internal auditory canal, but the otic capsule is spared until advanced changes are present in the remainder of the petrous pyramid. The bone changes in the pyramid begin in areas best supplied with marrow tissue. Involvement of the labyrinthine capsule, when present, begins in the outer periosteal layer; the middle endochondral layer is more resistant and the greatest resistance is present in the endosteal layer, but with extensive involvement these three layers can no longer be distinguished. Some have attributed the sensorineural loss to platybasia and basilar impression with torsion of the VIIIth nerve.

High resolution computerized tomography may also be used for more detailed information (Fig. 5) (Lloyd *et al.*, 1980).

FIG. 4
Paget's disease. Hypocycloidal tomograms showing Pagetoid bone replacing most of the periosteal bone at the petrous pyramid but also involving some of the bone of the labyrinthine capsule.

FIG. 5
CT scan—Paget's disease. Axial view showing disease affecting the skull base (woven bone). (Reproduced by kind permission of the Managing Editor of the *British Journal of Radiology* and the authors.)

Serum calcium is usually normal in Paget's disease, but immobilization will cause greater bone resorption than formation and as a result hypercalcaemia and hypercalciuria can occur. Serum alkaline phosphatase activity is elevated in active disease and particularly so if it is widespread. Activity of this enzyme is related to bone formation by osteoblasts and probably also by osteocytes. Serum acid phosphatase is an index of osteoclastic activity and is often increased in Paget's disease, particularly when the alkaline phosphatase is quite high, but its measurement is of little diagnostic value. Urinary hydroxyproline is an amino-acid found exclusively in collagen and in Paget's disease it may be greatly elevated when the condition is active, reflecting the breakdown of bone collagen.

More recently treatment has been attempted with synthetic human calcitonin by Menzies *et al.* (1975) and by Shai *et al.* (1971) with porcine calcitonin, and Grimaldi *et al.* (1975) with salmon calcitonin. Whilst such treatment has been associated with a striking reduction in the turnover of diseased bone confirmed by biochemical changes, there has been no significant hearing improvement in any of the patients. Walker *et al.* (1979) reported a follow-up of 3 years on the 13 patients treated earlier by the same group (Solomon *et al.*, 1975), but similarly reported no discernable difference between the treated and untreated groups.

Disodium etidronate (EHDP) is a diphosphonate which seems to possess all the biological properties of pyrophosphate including the ability to inhibit bone resorption. The drug appears to inactivate osteoclasts and osteoblasts and these properties have led to its trial in Paget's disease. However, long-term administration may result in histological osteomalacia associated with pathological fractures. It has the advantage that it may be taken orally but so far only one short report of its use in the deafness of Paget's disease has been published. Five patients were treated and their pure tone audiograms showed a significant improvement in the air conduction threshold of greater than 15 db. in 3 out of the 5 (Gennari and Sensini, 1975).

Trials have also been carried out designed to stimulate endogenous calcitonin secretion and to inhibit parathyroid-hormone secretion using a low phosphorus diet, aluminium hydroxide to further inhibit intestinal phosphorus absorption, calcium supplements between meals, and a thiazide diuretic to lower urinary calcium excretion.

Fibrous dysplasia

For many years, fibrous dysplasia of bone was not distinguished from primary hyperparathyroidism, and both kinds of osseous lesions were described pathologically and radiologically as osteitis fibrosa cystica.

Three types are now described:

Type I Monostotic—limited to one bone (usually femur, tibia, ribs or facial bones—particularly mandible and maxilla).

Type II Polyostotic (monomelic)—when more than one bone is involved (most frequently the lower limbs). In the skull the lesser and greater wings of the sphenoids and the vertical and horizontal processes of the frontal bones are mainly affected. The frontal and sphenoid sinuses are frequently obliterated.

Type III Disseminated with extraskeletal manifestations (McCune-Albright syndrome); bone distribution is similar to the polyostotic but commonly unilateral, with skin hyperpigmentations and endocrine disturbances.

Barrionuevo *et al.* (1980) found that the temporal bone was affected in 23 recorded cases of the monostotic and in 4 of the polyostotic form; so far there has been no report of the temporal bone being affected in Type III. They added 3 cases of their own (2 monostotic and 1 polyostotic).

The most common symptoms are loss of hearing and increased volume of the temporal region, mostly post-auricular. The hearing loss is caused by partial or complete obstruction of the external auditory canal, and direct or indirect involvement of the middle ear by fibrosseous proliferation (Fig. 6). When tinnitus was the first symptom, the disease affected the middle ear.

FIG. 6
Fibrous dysplasia. Dense bone obliterating the external auditory meatus on the left side.

Two cases have shown sensorineural hearing loss, one due to encroachment on the internal auditory canal, and the other due to a labyrinthine fistula (Chatterji, 1974; Cohen and Rosenwasser, 1969, respectively).

Serum calcium, phosphorus, and alkaline phosphatase have been found to be normal in all cases. Both the polyostotic and monostotic lesions are pathologically and radiologically identical. The radiological appearance of the disease is a function of its histological structure. A predominance of osseous elements renders the lesion more opaque, whilst a mixture of fibrous and bony elements produces a ground glass appearance, while predominance of fibrous elements produces a radiolucent cyst-like picture. Malignant change and the possible association between fibrous dysplasia and primary hyperparathyroidism have been reviewed by Williams and Thomas (1975). They also reported three cases.

Recently it has been suggested that primary hyperparathyroidism and fibrous dysplasia may occasionally occur together and that the serum calcium may occasionally be elevated in both the polyostotic form and the disseminated form of fibrous dysplasia, but never in the monostotic unless there is concomitant primary hyperparathyroidism. The serum phosphorus level is always normal. The alkaline phosphatase level is said to be raised in all types of fibrous dysplasia with the presence of an active lesion and it is always raised in the disseminated type. It would seem that the level of alkaline phosphatase is an indication of the activity of the lesion.

Leontiasis Ossea

The term Leontiasis was originally applied to describe the facial changes and distortion in leprosy. Later Virchow suggested the term Leontiasis Ossea to describe certain cases of hyperostosis of the skull (Knaggs, 1924). More recently it has been realized that under this descriptive term referring to enlargement of facial bones, most cases that have been reported are due to some recognized diseased entity, most commonly fibrous displasia and Paget's disease of bone (Byers and Jones, 1969).

Osteoscleroses (Osteopetrosis)

This comprises the two variants of osteopetrosis and pycnodystosis (Toulouse-Lautrec syndrome).

Osteopetrosis exists in two forms; the autosomal, dominant (benign), otherwise known as Albers-Schönberg's disease; and the autosomal recessive (malignant) type. Pathologically there is excessive formation of immature bone which results in thickening of the cortex and narrowing or even obliteration of the medullary cavity. In normal endochondral bone there is gradual transformation of immature bone into mature bone, whereas in osteopetrosis this transformation either does not occur or at a much reduced rate. This results in the formation of thick, dense and brittle bones ('chalk bones'). The abnormal development and reduced capacity of marrow cavities in diploic and diaphysial bones result in anaemia and extramedullary haematopoiesis. When remodelling of bone involves the cranial foramina, then stenosis and compression of emergent nerves and vessels may occur (Myers and Stool, 1969; Hamersma, 1970).

In the temporal bone, as elsewhere, it is the endochondral layer which is most severely affected and the entire mastoid air cell system may be absent, and filled instead with sclerotic bone. Myers and Stool (1969) found no inner ear abnormalities which could be attributed directly to the abnormal bone; likewise the internal auditory meatus may be narrowed, but the otic capsule remains unaffected (Hawke *et al.*, 1981).

In Albers-Schönberg disease, the hearing loss is predominantly conductive but may occasionally be mixed. In the malignant recessive

form, the hearing loss is sensorineural. In either form a facial palsy may occur or be the presenting sign. Successful decompression of the facial nerve has been carried out in this condition (Yarington and Sprinkle, 1967).

Serum calcium, phosphorus and alkaline phosphatase levels are normal, but the acid phosphatase may be markedly elevated; the urinary hydroxyproline levels are usually within normal limits (Johnston *et al.*, 1968).

Hyperostosis Corticalis Generalisata (van Buchem's disease)

This was first described by van Buchem *et al.* in 1955; further reports have appeared since (van Buchem *et al.*, 1962; van Buchem, 1971). This is an autosomal recessive condition, associated with normal stature and gross overgrowth of bone in the skull and skeleton, associated with facial palsy and conductive deafness, but normal digits (Fig. 7).

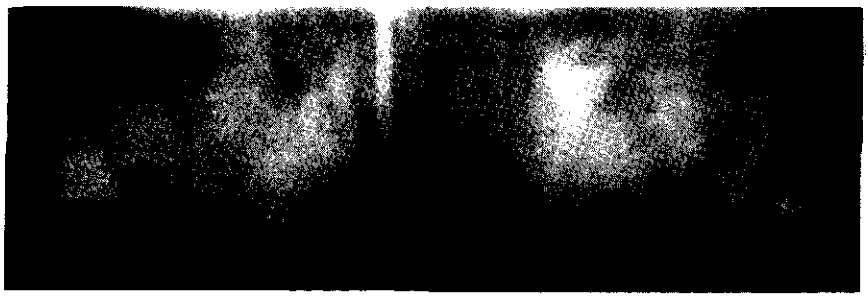

FIG. 7
van Buchem's disease. Sclerotic bone affecting both petrous pyramids with some tilt due to platybasia and bone softening.

There is osteosclerosis of the skull, mandible, clavicles and ribs, and hyperplasia of the diaphyseal cortex of the long and short bones. The skull and mandible may enlarge from the age of 10 onwards, with thickening of the calvaria, the skull base becoming dense, and with thickening of the clavicles, which thereby become palpable (Fig. 8). The facial paralysis may be unilateral or bilateral, and a gradually symmetrical hearing loss may be noted from the age of about 15. In some a sensorineural, in others a mixed loss may occur. Optic nerve involvement is a late complication. The serum calcium and phosphorus remain normal, but the alkaline phosphatase is frequently raised by as much as 50–250 per cent.

Sclerosteosis

This is an autosomal recessive condition in which skeletal overgrowth is associated with syndactyly and digital malformation; facial palsy and deafness are common complications and raised intracranial pressure may develop (Truswell, 1958).

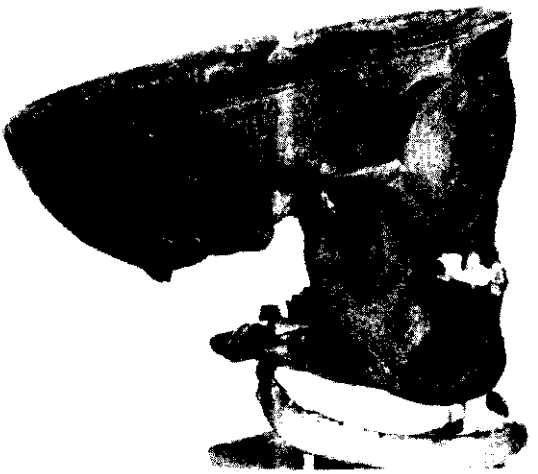

FIG. 8

Hyperostosis corticalis generalisata. Skull (right lateral view) and a rib; both external meatus grossly narrowed (internal meatus similarly affected). Girl, aged 10. 'Skull shows extraordinary degree of thickening and sclerosis of bones of calvarium . . ., obliteration of antral cavities.' Patient had bilateral nerve deafness and advanced optic atrophy (blind left eye). RCS Museum S.67.7 (Halliday, 1949).

The hearing loss may be bilateral sensorineural, mixed, or conductive. Facial nerve paralysis is often unilateral in childhood, becoming bilateral in late adolescence. There is also decreased sensory function of the ophthalmic and maxillary divisions of the Vth cranial nerve, anosmia and chronic headache.

The serum calcium, phosphorus and acid phosphatase concentrations are within normal limits, but the alkaline phosphatase is markedly elevated in nearly all patients. Radiologically the bones show increased density but abnormalities of bony modelling, if present, are of minor degree (Beighton *et al.*, 1976a and b).

Congenital Hyperphosphatasia (Osteoectasia)

This is a rare autosomal recessive condition with skeletal deformities developing in the second or third year of life. It is associated with dwarfing, fractures, and blue sclerae. There is marked irregular thickening of the skull and enlargement of the calvaria. The external auditory meatus may become narrowed and there is a progresive mixed hearing loss, which becomes evident from the fourth to the fourteenth year (approximately 60 to 80 db.). The serum alkaline and acid phosphatase are both consistently elevated.

Progressive Diaphyseal Dysplasia (Camurati-Engelmann's Disease; Osteopathia Hyperostotica Sclerositans Multiplex Infantilis)

This is an autosomal dominant condition principally involving the long bones, but the skull may be mildly affected. Generalized sclerosis of the

base similar to osteopetrosis may be seen but the vault bones are less commonly and less severely affected (Fig. 9).

Sparkes and Graham (1972) have reported the case of a 26-year-old man with progressive hearing difficulty leading to total deafness on the right side associated with a facial paralysis. Bilateral decompression of the slit like internal auditory canals was carried out and some initial improvement was noted.

Recently two cases have been described who underwent surgery (Miyamoto *et al.*, 1980), the first in a 26-year-old man who complained of a bilateral hearing loss, right-sided facial paralysis and chronic unsteadiness. X-rays showed bilateral massive overgrowth of dense bone involving the petrous apex and mastoid bone. Both internal auditory canals were partially obliterated by this dense bone; the second case was of a 30-year-old woman with bilateral sensorineural hearing loss, occurring suddenly 14 months earlier on the right side, and 9 months later on the left. Both cases were explored surgically by a middle cranial fossa approach, the first to improve the facial nerve function and the second to decompress the internal auditory canal on the right side. Following surgery the hearing of the second patient has remained stable and further X-rays did not show evidence of recompression.

Neurofibromatosis

This is a common autosomal dominant disorder of neural tissue, described by von Recklinghausen in 1882. It is characterized by multiple pale brown patches on the skin, regular in outline (soft fibromata on or

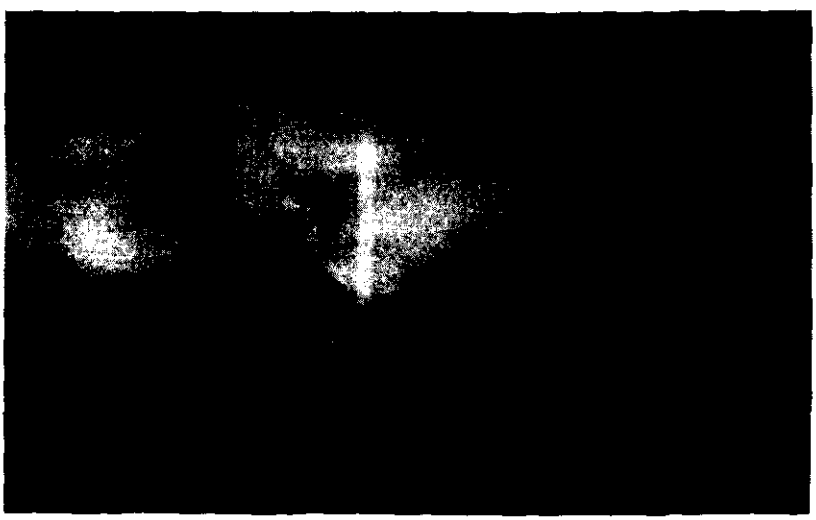

FIG. 9
Engelmann's disease. Dense bone appears to be narrowing the internal auditory meatus.

FIG. 10
Neurofibromatosis. Frontal view of skull of J.M., aged 29.

FIG. 11
Neurofibromatosis. Right lateral view of skull of J.M., aged 29.

within the skin), and tumours arising from nerves, e.g. acoustic neuromata. Bone lesions occur in about half the cases (Hunt and Pugh, 1961; Nordin, 1973; Beighton, 1978). Figs. 10 and 11 show the famous patient of Sir Frederick Treves, Joseph Merrick. From these it will be seen that he has marked narrowing of the right external auditory meatus, but there is no evidence that amongst his misfortunes, he suffered from an acoustic neuroma (Fig. 12). (He died at the age of 29.)

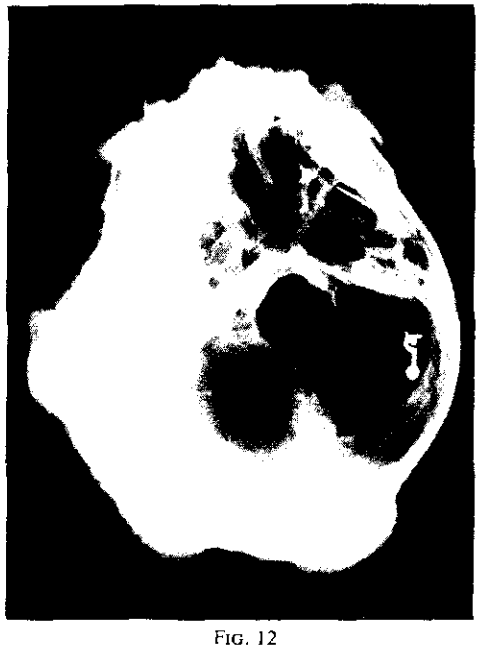

FIG. 12
X-ray. Towne's view of skull. J.M., aged 29.

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J. B. Booth, F.R.C.S.,
The London Hospital,
Whitechapel,
London, E1 1BB.